14. MALFORMATIONS OF THE UROGENITAL TRACT (L)

In view of the common origin of large parts of the urinary and genital systems from the urogenital ridge it is not surprising that malformations of the urinary and genital tract occur together at least as frequently as separately. In many instances it is semasiological to discuss to which system a specific malformation belongs, as in hypospadias or a vesico-vaginal fistula, and, as already noted in section 7, defects of differentiation from a common cloaca determine many defects of the genitalia and urinary tract in common with those of the gut.

In addition, for reasons not explicable on a basis of development from the same rudiments, urogenital malformations are often associated with malformations elsewhere in the body, notably of the ear, the heart and great vessels and the extremities. Failures of fusion of the face and the palate and exomphalos are also associated in some infants. Malformations of the urogenital tract are also common in the B group—notably the association of anencephalus with horseshoe kidney—and some cases in the E2 group could logically have been included in the L group.

In Table 14.1 an attempt has been made to group these malformations in the L group into four categories which are by no means satisfactory but may have certain advantages. References are also given to cases where there are urogenital and other anomalies in infants classed in the multiple or N group. It proved extremely difficult, on occasion, to decide into which group to place many cases.

PSEUDOHERMAPHRODITISM

In the conditions listed in the first numbered column of Table 14.1 there has always been mention of ambiguity of genitalia or severe perineal hypospadias. The latter merges into the former clinically and these cases in total represent what is loosely called pseudohermaphroditism. Such cases could only be classified adequately after careful study of the morbid anatomy, the histology of the gonads, chromosomal and other studies. The frequency of this group is about 0.12 per 1000 single births. However, as will be noted from other sections and the listed malformations in the Basic Tabulations by Centres booklet, there are many cases of "indeterminate sex" or "pseudohermaphroditism" in the multiple or N group, the E2 group and the B group.

MALFORMATIONS OF THE KIDNEY AND URINARY TRACT

In the second numbered column of Table 14.1 are included all the specific malformations of the kidney, such as horseshoe kidney, agenesis, ectopia, duplications of parts of the tract and the ureteral and urethral stenoses. All simple hydronephroses are there included. There must be many more such malformations which permit survival and are manifest only in later life.

CYSTIC KIDNEYS

All the cases where only cystic kidneys are mentioned are in column 3 of Table 14.1. From the descriptions, the malformations reported were all of the many small-cyst types. The histopathology of these conditions is very complex and it is impossible to attempt to classify on the basis of the information given. None of the very large cystic kidney types appears to have been encountered in the study.

It is important to remember that these reported cases must represent a small proportion only of congenital cystic anomalies. In the first place they are only identifiable in cases examined at autopsy. In the second place, by no means all cases are fatal at birth. Thirdly, it has to be remembered that there are several types caused by dominant and by recessive gene mutations which cause trouble only later in life although the developmental anomalies must have been present at birth.

OTHER AND MULTIPLE UROGENITAL MALFORMATIONS

In column 4 of Table 14.1 are included all other types of urogenital malformation, such as uterus didelphys, extraversion of the bladder and the complex cases where anomalies of the external genitalia are associated with defects of the internal genitalia or the urinary tract.

Only consultation of the listed malformations under M and N in the Basic Tabulations by Centres booklet will enable the recorded range to be appreci-

ated. Only careful anatomical, endocrinological, family and chromosomal studies could reveal the true and enormous range of these disorders.

UROGENITAL TRACT MALFORMATIONS OCCURRING IN THE N GROUP

In all, there were 107 cases where the anomalies were all of the urogenital tract (Table 14.1). However, there were also 102 cases with such defects in the N group (69 males and 24 females and 9 of indeterminate sex). There seems little doubt that in fact only in a minority of cases is only the urogenital tract affected, because more autopsies would reveal anomalies of other systems in those presently classed in the L group and more urogenital anomalies in cases in several other groups.

UROGENITAL TRACT MALFORMATIONS IN THE " MINOR " GROUP

In the " minor " group there were 244 cases of hypospadias (some of these probably required operative treatment), 10 cases of epispadias (which may all have needed treatment) and 134 cases of congenital hydrocoele.

It seems likely that the over-all frequency of occurrence of some defect of the urogenital tract must be about 1 in 1000 total births.

DISCUSSION

Apart from one specified case of Turner's syndrome there is little information on which to base opinions as to the contribution of gene mutations or chromosomal aneuploidy to the cases reported. This is not surprising as few cases of Turner's syndrome, XXX " super-females " or XXY Klinefelter males would be recognized at birth. The sex-linked (or sex-limited) testicular feminization syndromes and the autosomal recessive syndromes determining ovarian dysgenesis would not be recognized.

Perhaps of the gene mutation syndromes only adrenal virilization would be recognized at birth and of the syndromes associated with chromosomal abnormalities that small number of true hermaphrodites where the karyotype is not simple XX and a variety of individually exceedingly uncommon cases with sex chromosome mosaicism or structural changes. In one case (Madrid, Group N, No. 7) the infant appeared to have been a true hermaphrodite as there was an ovary on one side and a testis on the other.

The great majority of cases, however, appear to fall into the category of " phenodeviants " (see section 20), i.e., aberrant development, in this case of all the structures developed from the urogenital ridge, there being no simple genetical hypothesis to explain any genetical contribution to the etiology.

TABLE 14.1
UROGENITAL TRACT MALFORMATIONS (L) IN SINGLE BIRTHS

CENTRE		Number of cases																				References to N group cases (see malformations listed in Basic Tabulations by Centres Booklet)
		1 Indeterminate or ambiguous sex				2 Malformations of urinary tract only				3 Cystic kidneys				4 Other and combinations of (1 - 3)				Total				
		M	F	NR	T	M	F	NR	T	M	F	NR	T	M	F	NR	T	M	F	NR	T	
I 1	MELBOURNE	1	-	-	1	5	-	-	5	-	-	-	-	-	-	-	-	6	-	-	6	N 1, 4, 5, 12, 13
I 2	MELBOURNE	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	N 1, 2, 4, 8
II	SAO PAULO	2	1	3	6	-	2	-	2	-	2	-	2	-	-	-	-	2	5	3	10	N 2, 3, 7
III	SANTIAGO	1	3	2	6	-	-	-	-	-	-	-	-	-	-	-	-	1	3	2	6	N 1, 2
IV 1	BOGOTA	2	-	-	2	2	1	-	3	-	-	-	-	-	-	-	-	4	1	-	5	N 1, 3, 4, 10
IV 2	MEDELLIN	1	-	1	2	-	1	-	1	-	-	-	-	-	-	-	-	1	1	1	3	N 4, 5, 12, 20
V	CZECHOSLOVAKIA	5	-	1	6	-	2	-	2	-	-	-	-	2	1	-	3	7	3	1	11	N 1, 3, 5, 6, 9, 10, 12, 13, 14 18, 19, 21, 22, 23, 25, 31
VI	ALEXANDRIA	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	N 3, 5
VII	HONG KONG	-	1	-	1	-	-	-	-	-	-	-	-	-	1	-	1	-	2	-	2	N 1, 4, 8, 14
VIII 1	BOMBAY	-	1	-	1	-	-	-	-	1	1	-	2	3	-	-	3	4	2	-	6	N 2, 6, 15, 18
VIII 2	CALCUTTA	2	-	2	4	-	-	-	-	-	-	-	-	-	-	-	-	2	-	2	4	
IX 1	KUALA LUMPUR	-	-	-	-	3	-	-	3	-	-	-	-	-	-	-	-	3	-	-	3	N 8
IX 2	SINGAPORE	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	N 6, 7, 9, 10, 11, 12, 13
X 1	MEXICO CITY	1	-	-	1	-	-	-	-	2	-	-	2	1	3	-	4	4	3	-	7	N 1, 11, 15, 16, 18, 21, 22, 26
X 2	MEXICO CITY	-	-	1	1	-	-	-	-	1	-	-	1	-	3	-	3	1	3	1	5	N 8, 10
XI	BELFAST	3	1	-	4	-	-	-	-	-	2	-	2	-	-	-	-	3	3	-	6	N 9, 14, 15
XII	PANAMA CITY	1	-	1	2	-	-	-	-	-	-	-	-	-	-	-	-	1	-	1	2	N 2, 5, 10, 11, 12
XIII	MANILA	3	1	-	4	-	-	-	-	-	-	-	-	-	1	-	1	3	2	-	5	N 1, 6, 15, 16, 18, 24, 29
XIV 1	CAPE TOWN	-	-	-	-	-	-	-	-	-	-	-	-	1	-	-	1	1	-	-	1	
XIV 2	JOHANNESBURG	5	-	-	5	-	2	-	2	-	-	-	-	-	2	-	2	5	4	-	9	N 3, 4, 6, 8, 9, 11, 12, 14, 17, 18
XIV 3	PRETORIA	1	-	1	2	-	-	-	-	-	-	-	-	-	-	-	-	1	-	1	2	
XV	MADRID	1	1	-	2	4	-	-	4	-	-	-	-	-	1	-	1	5	2	-	7	N 1, 2, 3, 4, 5, 6, 7, 8
XVI 1	LJUBLJANA	-	-	-	-	-	1	-	1	-	1	-	1	-	-	-	-	-	2	-	2	N 4, 7
XVI 2	ZAGREB	2	-	-	2	-	-	-	-	1	1	-	2	-	1	-	1	3	2	-	5	N 6
	TOTAL	31	9	12	52	14	9	-	23	5	7	-	12	7	13	-	20	57	38	12	107	(102)